downward palpebral fissures, low-set ears rotated backward, triangular face, micrognathia, high-arched palate, widely spaced nipples, cardiac operation scar, and pectus excavatum. Full blood count, biochemical analysis, and thyroid function tests were found to be normal in terms of short stature.

Since the dysmorphic features were consistent with Noonan syndrome, p.D61G heterozygote mutation in *PTPN11* gene was found. The patient with Noonan syndrome was started treatment with 45 micrograms/kg/dose of growth hormone because of short stature and insufficient height velocity.

In this report, Noonan syndrome patient associated with heterozygous P.D61G mutation was presented. Early diagnosis and appropriate treatment will prevent the development of complications.

(P-12)

A New Mutation in an Infant with Hypercalcemia

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Hypercalcemia is a serious condition, which may threaten life. Familial benign hypocalciuric hypercalcemia (FBHH) is a benign situation which develops due to inactivating mutation of calcium sensing receptor (CaSR). Differential diagnosis of FBHH is important in order to prevent unnecessary laboratory tests and treatments.

A three-month-old infant, who was born 2950 g on 38th gestational week to healthy unrelated parents, was admitted to pediatric endocrinology department due to high TSH levels. Her anthropometric evaluation was appropriate for her age and she did not have any dysmorphic stigmata. She was diagnosed with hypothyroidism and put on LT4 treatment. When she was 5 months old, her serum calcium was 11.34 mg/dL (8.8-10.6).

The patient did not have any clinical symptoms of hypercalcemia. Serum phosphorus, parathormone, vitamin levels, urine calcium/creatinine, urine analysis, and renal ultrasonography were normal. Oral hydration was started and her calcium level decreased to 10.8 mg/dL on follow-up. Biochemical and hormonal parameters of father and mother of the patient were evaluated to determine the etiology of hypercalcemia. Whole exome sequencing was performed to the patient and homozygote mutation on exon 7 (p.Glu1011Gln/c.3031G>C) was detected. In order to determine if the parents of the patient had heterozygote mutation or if this is a *de novo* mutation, genetic analysis was also performed to the parents.

To the best of our knowledge, the mutation found in our patient was not mentioned in the literature before. We think that this mutation may cause FBHH in our case. Functional evaluation should be performed for definitive diagnosis.

(P-13)

An Infant with Leydig Cell Hypoplasia Presenting with Bilateral Inguinal Masses

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Luteinizing hormone/chorionic gonadotropin receptor *(LHCGR)* is essential for normal male sex differentiation. Inactivating mutations of *LHCGR* gene result in varying degree of Leydig cell hypoplasia (LCH) that causes 46,XY DSD.

A 2-year and 1-month-old female infant was referred to us for further evaluation of DSD. She was the fourth child of healthy consanguineous Turkish parents and was born at 38 weeks. The patient was raised as female. Her mother noticed bilateral masses on her inguinal areas and brought her to the local hospital. The abdomen USG revealed bilateral masses (possibly testicular structures) on both inguinal region. On admission, her weight was 0.95 SDS, height was 3.4 SDS, and physical examination was normal except for the palpable gonads in both inguinal regions. Her external genitalia was completely female in appearance.

Hormonal investigations showed low testosterone (13.6 ng/mL) with high gonadotropin (follicle-stimulating hormone = 6.1 IU/L and LH = 12.53 IU/L) and AMH (>23 ng/mL) levels. Serum levels of 17-OHP, DHEAS, and AS were within normal ranges. Testosterone response to 3-day HCG stimulation test was absent (sT = 14.47 ng/dL). The karyotype was 46,XY. Pelvic ultrasound revealed absent uterus and ovaries but presence of testicular structures in the superior inguinal canal bilaterally. Bone age was 2 years. The diagnosis of LCH was considered in the patient. *LHCGR* gene sequencing demonstrated a homozygous c.1435C>T (p.R479*) mutation that confirmed the diagnosis. In the parents genetic analysis is being done.

Although LHC is usually diagnosed at pubertal or postpubertal period, this case demonstrates that LCH can be seen in infancy period presenting with inguinal masses.